# ST3GAL5 Gene

Subjects: Genetics & Heredity
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ST3 beta-galactoside alpha-2,3-sialyltransferase 5

genes

## 1. Normal Function

The *ST3GAL5* gene provides instructions for making an enzyme called GM3 synthase. This enzyme carries out a chemical reaction that is the first step in the production of certain fatty molecules (lipids) called gangliosides. Specifically, GM3 synthase converts a molecule called lactosylceramide to a simple ganglioside called GM3. Further reactions use GM3 to create more complex gangliosides.

Gangliosides are present on the surface of cells and tissues throughout the body, and they are particularly abundant in the nervous system. Although their exact functions are unclear, studies suggest that these molecules help regulate chemical signaling pathways that influence cell growth and division (proliferation), cell movement (motility), the attachment of cells to one another (adhesion), and cell survival. Gangliosides appear to be important for normal brain development and function.

## 2. Health Conditions Related to Genetic Changes

### 2.1. GM3 synthase deficiency

At least one mutation in the *ST3GAL5* gene has been found to cause GM3 synthase deficiency, a condition characterized by recurrent seizures (epilepsy) and problems with brain development. The known mutation replaces a single protein building block (amino acid), arginine, with a signal to stop protein production prematurely. The mutation is written as Arg288Ter or R288X, although in older scientific articles it is sometimes written as Arg232Ter or R232X. The mutation prevents the production of any functional GM3 synthase. Without this enzyme, cells cannot produce GM3 or other gangliosides normally. It is unclear how a loss of this enzyme leads to the signs and symptoms of GM3 synthase deficiency. Researchers are working to determine whether it is the lack of gangliosides or a buildup of compounds used to make gangliosides, or both, that underlies the seizures and other problems with brain development that occur in this condition.

## 3. Other Names for This Gene

- alpha 2,3-sialyltransferase V
- CMP-NeuAc:lactosylceramide alpha-2,3-sialyltransferase
- ganglioside GM3 synthase
- GM3 synthase
- lactosylceramide alpha-2,3-sialyltransferase
- lactosylceramide alpha-2,3-sialyltransferase isoform 1
- lactosylceramide alpha-2,3-sialyltransferase isoform 2
- SATI
- sialyltransferase 9 (CMP-NeuAc:lactosylceramide alpha-2,3-sialyltransferase; GM3 synthase)
- SIAT9
- SIATGM3S
- ST3Gal V
- ST3GalV

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